



**WORKSHOP PRESENTATION**

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# Characterization of both myocardial extracellular volume expansion and myocyte hypertrophy by CMR detect early signs of myocardial tissue remodeling in Friedreich's ataxia patients without heart failure.

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## Background

Heart Failure (HF) is the most common cause of death in Friedreich's ataxia (FRDA), a mitochondrial disease characterized by neurodegeneration, hypertrophic cardiomyopathy, caused by homozygous GAA expansions in the *FXN* gene. Recent report demonstrates that specific-gene therapy may prevent and reverse the cardiomyopathy in a mice model of FRDA. Myocardial interstitial fibrosis is a hallmark of FRDA's cardiomyopathy and a potential substrate for arrhythmias and HF. Myocardial tissue characterization by cardiac magnetic resonance (CMR) allows access to tissue-based phenotypes that may better describe LV remodeling in FRDA's cardiomyopathy.

## Methods

The aim of this study was to perform direct quantification of myocardial extracellular volume fraction (ECV) and intracellular lifetime of water ( $\tau_{ic}$ ), a measure of cardiomyocyte hypertrophy, using T1-weighted CMR imaging in cohort of patients with FRDA without HF.

We investigated 27 FRDA patients without HF (mean age  $26.8 \pm 9$ , 12 female) and in 30 healthy volunteers as control subjects (mean age  $49 \pm 15$ ) using a 3T CMR system. The T1 quantification by Look-Locker gradient-echo before and after contrast applying a 2-site model for transcytolemmal water Exchange was used for ECV and  $\tau_{ic}$

quantification. Cine CMR and LGE imaging in matching locations were also performed.

## Results

FRDA patients revealed normal LVEF with increased LV Mass-index compared with health controls (for LVEF  $67.3\% \pm 11.5$  vs.  $62.5\% \pm 6.8$ ,  $P = \text{NS}$ ; for LVMASSi  $62.7 \pm 23$  vs.  $45.1 \pm 6.8 \text{ g/m}^2$ ,  $p < 0.05$ ). In 4 out 27 FRDA patients a non-ischemic LGE pattern was present. Both ECV and intracellular lifetime of water ( $\tau_{ic}$ ) were significantly higher FRDA patients (ECV:  $0.36 \pm 0.04$  vs.  $0.28 \pm 0.03$ ,  $p < 0.0001$ ;  $\tau_{ic}$ :  $0.12 \pm 0.08$  vs.  $0.08 \pm 0.03$ ,  $p < 0.005$ ).

## Conclusions

ECV and intracellular lifetime of water ( $\tau_{ic}$ ) determined by T1 measurements characterized early signs of myocardial tissue remodeling in FRDA with normal LVEF. Early changes in tissue-phenotypes are detectable by novel-CMR methods in FRDA patients, and may be useful to track effects of new genetic therapies for FRDA cardiomyopathy.

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